Galactosemia

Galactosemia is an inherited autosomal recessive disorder of carbohydrate metabolism. Classic galactosemia is due to a deficiency of the enzyme galactose-1-phosphate uridyl transferase, which leads to an accumulation of total galactose. In nature, galactose is combined with glucose to form lactose, the primary sugar in human milk and commercial (non-soy) infant formulas. Affected infants are not able to metabolize this causing the build up of galactose in the body, which can lead to cellular damage and even death. There are several benign genetic variants characterized by a less severe reduction in enzyme activity (e.g. Duarte variant). These children often present with a persistent positive newborn screen but are asymptomatic. They do not require treatment and remain clinically well on breast-milk and standard infant formulas.

Estimated Incidence:	1:42,000
Laboratory Screening Test:	Quantitative GALT enzyme done on all infants Quantitative fluorometric assay to detect Galactose + Gal-1- P (total galactose) done on all positive tests and transfused infants.
Timing of Test:	Valid at birth
Feeding Effect:	None. Enzyme activity not affected by feeding
Transfusion Effect:	Transfusion of red blood cells may interfere with the accuracy of testing causing a false negative result. **Obtain newborn screen before transfusion.**
Confirmation:	All strong and persistent borderline positive tests are referred to the Pediatric Neurology metabolic Clinic (PNMC) for confirmation (734) 763-4697. **Do not send diagnostic labs before contacting the PNMC.**
Treatment:	Immediate change to soy formula and lifelong exclusion of galactose from the diet.

For further information, contact the Newborn Screening Program
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